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Mouse Sox9 Sequence

AGTTTCAGTC CAGGAACTTT TCTTTGCAAG AGAGACGAGG TGCAAGTGGC CCCGGTTTCG TTCTCTGTTT TCCCTCCCTC CTCCTCCGCT CCGACTCGCC 51 101 TTCCCCGGGT TTAGAGCCGG CAGCTGAGAC CCGCCACCCA GCGCCTCTGC TAAGTGCCCG CCGCCGCAGC CCGGTGACGC GCCAACCTCC CCGGGAGCCG 151 TTCGCTCGGC GTCCGCGTCC GGGCAGCTGA GGGAAGAGGA GCCCCAGCCG 201 CCGCGGCTTC TCGCCTTTCC CGGCCACCCG CCCCTGCCC CGGGCTCGCG 251 301 TATGAATCTC CTGGACCCCT TCATGAAGAT GACCGACGAG CAGGAGAAGG GCCTGTCTGG CGCCCCCAGC CCCACCATGT CGGAGGACTC GGCTGGTTCG 351 401 CCCTGTCCCT CGGGCTCCGG CTCGGACACG GAGAACACCC GGCCCCAGGA 451 GAACACCTTC CCCAAGGGCG AGCCGGATCT GAAGAAGGAG AGCGAGGAAG 501 ATAAGTTCCC CGTGTGCATC CGCGAGGCGG TCAGCCAGGT GCTGAAGGGC TACGACTGGA CGCTGGTGCC CATGCCCGTG CGCGTCAACG GCTCCAGCAA 551 GAACAAGCCA CACGTCAAGC GACCCATGAA CGCCTTCATG GTGTGGGCGC 601 651 AGGCTGCGCG CAGGAAGCTG GCAGACCAGT ACCCGCATCT GCACAACGCG 701 GAGCTCAGCA AGACTCTGGG CAAGCTCTGG AGGCTGCTGA ACGAGAGCGA 751 GAAGAGCCC TTCGTGGAGG AGGCGGAGCG GCTGCGCGTG CAGCACAAGA 801 AAGACCACCC CGATTACAAG TACCAGCCCC GGCGGAGGAA GTCGGTGAAG 851 AACGGACAAG CGGAGGCCGA AGAGGCCACG GAACAGACTC ACATCTCTCC 901 TAATGCTATC TTCAAGGCGC TGCAAGCCGA CTCCCCACAT TCCTCCTCCG GCATGAGTGA GGTGCACTCC CCGGGCGAGC ACTCTGGGCA ATCTCAGGGT 951 1001 CCGCCGACCC CACCCACCAC TCCCAAAACC GACGTGCAAG CTGGCAAAGT 1051 TGATCTGAAG CGAGAGGGGC GCCCTCTGGC AGAGGGGGGC AGACAGCCCC 1101 CCATCGACTT CCGCGACGTG GACATCGGTG AACTGAGCAG CGACGTCATC TCCAACATTG AGACCTTCGA CGTCAATGAG TTTGACCAAT ACTTGCCACC 1151 CAACGGCCAC CCAGGGGTTC CGGCCACCCA CGGCCAGGTC ACCTACACTG 1201 GCAGTTACGG CATCAGCAGC ACCGCACCCA CCCTGCGAC CGCGGGCCAC 1251

Figure 1(a)

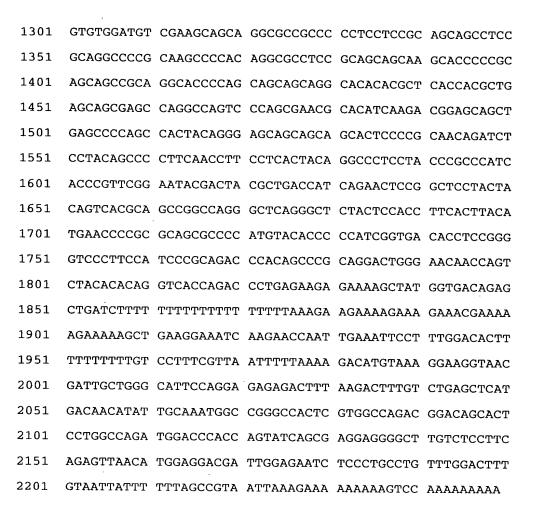


Figure 1(b)

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Mouse Sox-9 amino acid sequence

Met Asn Leu Leu Asp Pro Phe Met Lys Met Thr Asp Glu Gln Glu Lys Gly Leu Ser Gly Ala Pro Ser Pro Thr Met Ser Glu Asp Ser Ala Gly Ser Pro Cys Pro Ser Gly Ser Gly Ser Asp Thr Glu Asn Thr Arg Pro Gln Glu Asn Thr Phe Pro Lys Gly Glu Pro Asp Leu Lys Lys Glu Ser Glu Glu Asp Lys Phe Pro Val Cys Ile Arg Glu Ala Val Ser Gln Val Leu Lys Gly Tyr Asp Trp Thr Leu Val Pro Met Pro Val Arg Val Asn Gly Ser Ser Lys Asn Lys Pro His Val Lys Arg Pro Met Asn Ala Phe Met Val Trp Ala Gln Ala Ala Arg Arg Lys Leu Ala Asp Gln Tyr Pro His Leu His Asn Ala Glu Leu Ser Lys Thr Leu Gly Lys Leu Trp Arg Leu Leu Asn Glu Ser Glu Lys Arg Pro Phe Val Glu Glu Ala Glu Arg Leu Arg Val Gln His Lys Lys Asp His Pro Asp Tyr Lys Tyr Gln Pro Arg Arg Arg Lys Ser Val Lys Asn Gly Gln Ala Glu Ala Glu Glu Ala 185 Thr Glu Gln Thr His Ile Ser Pro Asn Ala Ile Phe Lys Ala Leu Gln Ala Asp Ser Pro His Ser Ser Ser Gly Met Ser Glu Val His Ser Pro Gly Glu His Ser Gly Gln Ser Gln Gly Pro Pro Thr Pro Pro Thr Thr 230 235 Pro Lys Thr Asp Val Gln Ala Gly Lys Val Asp Leu Lys Arg Glu Gly 250 Arg Pro Leu Ala Glu Gly Gly Arg Gln Pro Pro Ile Asp Phe Arg Asp

Figure 1(c)

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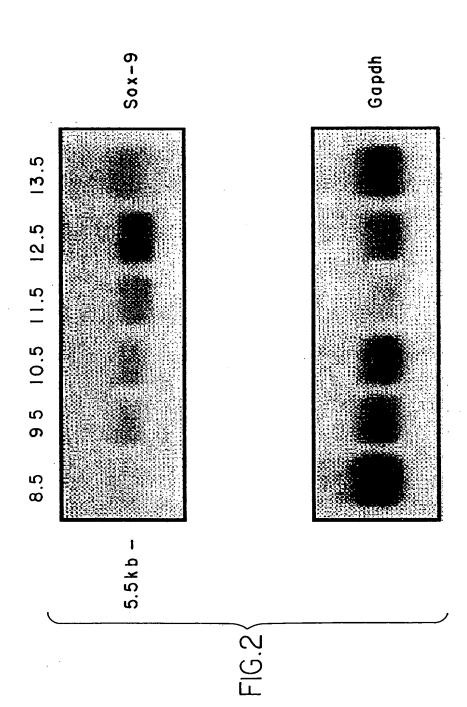
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Val Asp Ile Gly Glu Leu Ser Ser Asp Val Ile Ser Asn Ile Glu Thr Phe Asp Val Asn Glu Phe Asp Gln Tyr Leu Pro Pro Asn Gly His Pro Gly Val Pro Ala Thr His Gly Gln Val Thr Tyr Thr Gly Ser Tyr Gly 310 Ile Ser Ser Thr Ala Pro Thr Pro Ala Thr Ala Gly His Val Trp Met Ser Lys Gln Gln Ala Pro Pro Pro Pro Gln Gln Pro Pro Gln Ala Pro Gln Ala Pro Gln Ala Pro Pro Gln Gln Gln Ala Pro Pro Gln Gln 360 Pro Gln Ala Pro Gln Gln Gln Ala His Thr Leu Thr Thr Leu Ser Ser Glu Pro Gly Gln Ser Gln Arg Thr His Ile Lys Thr Glu Gln Leu 390 Ser Pro Ser His Tyr Arg Glu Gln Gln Gln His Ser Pro Gln Gln Ile Ser Tyr Ser Pro Phe Asn Leu Pro His Tyr Arg Pro Ser Tyr Pro Pro 425 Ile Thr Arg Ser Glu Tyr Asp Tyr Ala Asp His Gln Asn Ser Gly Ser Tyr Tyr Ser His Ala Ala Gly Gln Gly Ser Gly Leu Tyr Ser Thr Phe Thr Tyr Met Asn Pro Ala Gln Arg Pro Met Tyr Thr Pro Ile Gly Asp 470 Thr Ser Gly Val Pro Ser Ile Pro Gln Thr His Ser Pro Gln Asp Trp 485 490 Glu Gln Pro Val Tyr Thr Gln Val Thr Arg Pro

Figure 1(d)





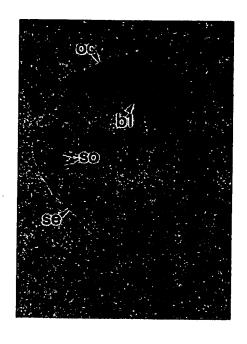


FIG.3a

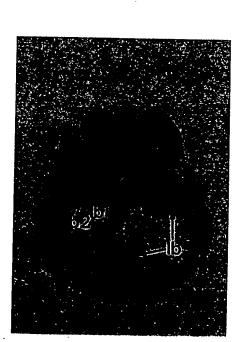


FIG.3c

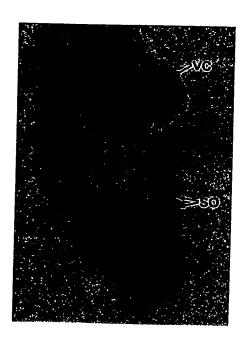


FIG.3b

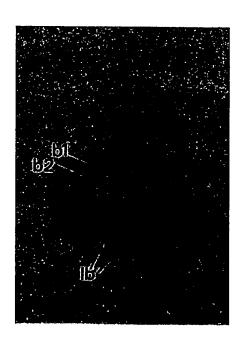


FIG.3d

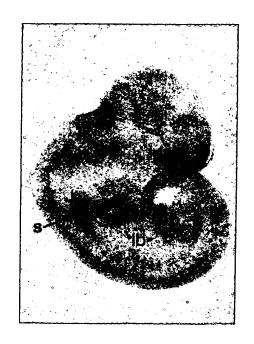


FIG.3e

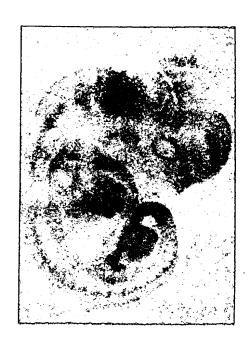


FIG.3f



FIG.3g

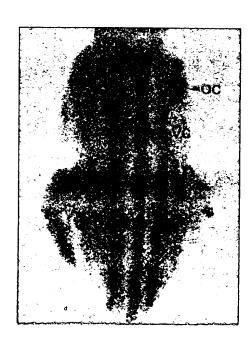


FIG.3h

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FIG.3i

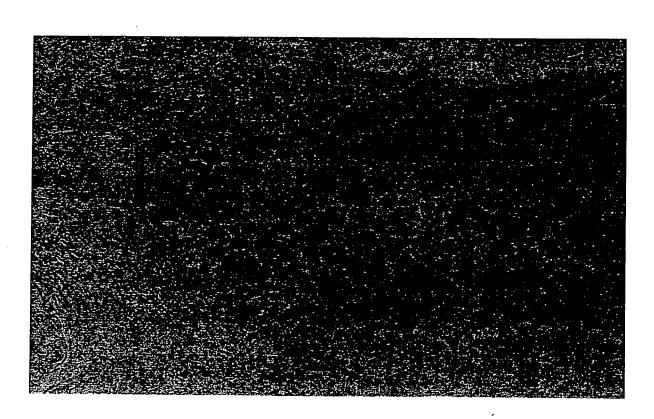


FIG.4

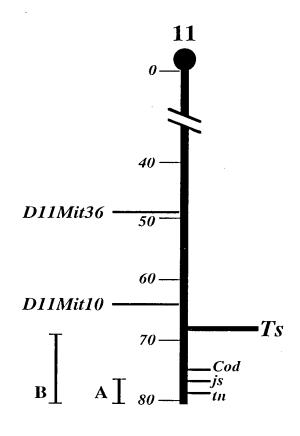
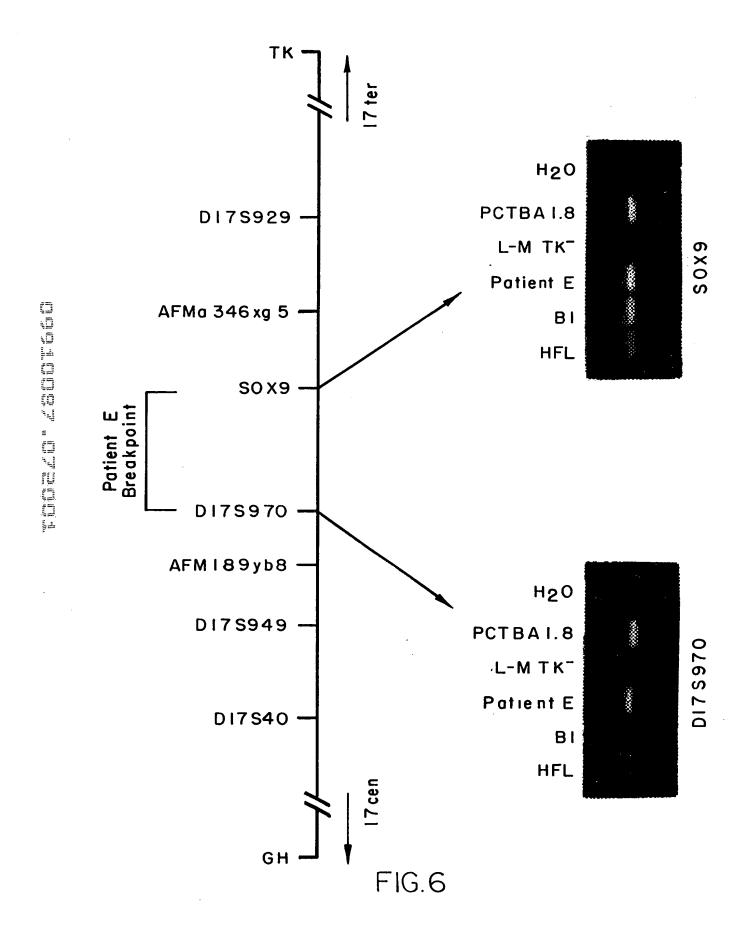
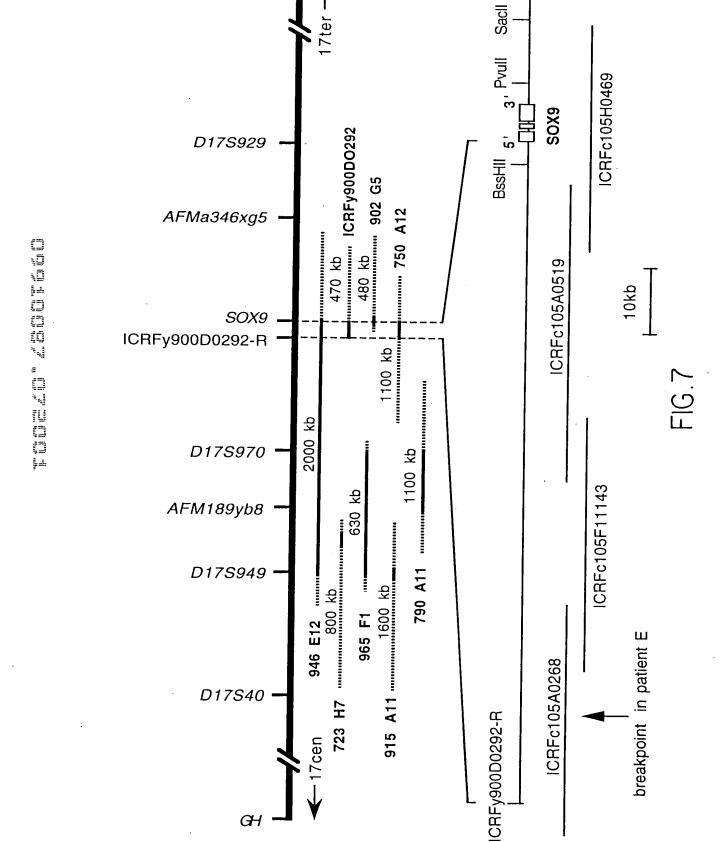


FIG.5

O.G. FIG.			
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BY	CLASS	SUBCLASS
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CGGAGCTCGA AACTGACTGG AAACTTCAGT GGCGCGGAGA CTCGCCAGTT TCAACCCCGG CGGAGCTCGA AACTGACTGG AAACTTCAGT GGCGCGGAGA CTCGCCAGTT TCAACCCCGG
AAACTTTTCT TTGCAGGAGG AGAAGAGAAG GGGTGCAAGC GCCCCACTT TTGCTCTTTT
TCCTCCCCTC CTCCTCCTCT CCAATTCGCC TCCCCCACT TGGAGCGGGC AGCTGTGAAC
TGGCCACCCC GCGCTTCCT AAGTGCTCGC CGCGGTAGCC GGCCGACGCG CCAGCTTCCC
CGGGAGCCGC TTGCTCCGCA TCCGGGCAGC CGAGGGGAGA GGAGCCCGCG CCTCGAGTCC
CCGAGCCGCC GCGCTTCTC GCCTTTCCCG GCCACCAGCC CCCTGCCCCG GGCCCGCGTA
TGAATCTCCT GGACCCCTTC ATGAAGATGA CCGACGAGCA GGAGAAGGGC CTGTCCGGCG
CCCCCAGCCC CACCATGTCC GAGGACTCCG CGGGCTCGCC CTGCCCGTCG GGCTCCGGCT
CGGACACCGA GAACACGCGG CCCCAGGAGA ACACGTTCCC CAAGGGCGAG CCCGATCTGA
AGAAGGAGAG CGAGGAGGAC AAGTTCCCCG TGTGCATCCG CGAGGCGGTC AGCCAGGTGC
TCAAAGGCTA CGACTGGACG CTGGTGCCCA TGCCGGTGC CGTCAACGGC TCCAGCAAGA TCAAAGGCTA CGACTGGACG CTGGTGCCCA TGCCGGTGCG CGTCAACGGC TCCAGCAAGA ACAAGCCGCA CGTCAAGCGG CCCATGAACG CCTTCATGGT GTGGGCGCAG GCGGCGCGCA GGAAGCTCGC GGACCAGTAC CCGCACTTGC ACAACGCCGA GCTCAGCAAG ACGCTGGGCA AGCTCTGGAG ACTTCTGAAC GAGAGCGAGA AGCGGCCCTT CGTGGAGGAG GCGGAGCGGC TGCGCGTGCA GCACAAGAAG GACCACCCGG ATTACAAGTA CCAGCCGCGG CGGAGGAAGT CGGTGAAGAA CGGGCAGGCG GAGGCAGAGG AGGCCACGGA GCAGACGCAC ATCTCCCCCA ACGCCATCTT CAAGGCGCTG CAGGCCGACT CGCCACACTC CTCCTCCGGC ATGAGCGAGG TGCACTCCCC CGGCGAGCAC TCGGGGCCAAT CCCAGGGCCC ACCGACCCCA CCCACCACCC CCAAAACCGA CGTGCAGCCG GGCAAGGCTG ACCTGAAGCG AGAGGGGCGC CCCTTGCCAG AGGGGGGCAG ACAGCCCCCT ATCGACTTCC GCGACGTGGA CATCGGCGAG CTGAGCAGCG ACGTCATCTC CAACATCGAG ACCTTCGATG TCAACGAGTT TGACCAGTAC CTGCCGCCCA ACGGCCACCC GGGGGTGCCG GCCACGCACG GCCAGGTCAC CTACACGGGC AGCTACGGCA TCAGCAGCAC CGCGGCCACC CCGGCGAGCG CGGGCCACGT GTGGATGTCC AAGCAGCAGG CGCCGCCGC ACCCCGCAG CAGCCCCCAC AGGCCCCGCC GGCCCCGCAG GCGCCCCCGC ACTACACCGA CCACCAGAAC TCCAGCTCCT ACTACAGCCA CGCGGCAGGC CAGGGCACCG GCCTCTACTC CACCTTCACC TACATGAACC CCGCTCAGCG CCCCATGTAC ACCCCCATCG CCGACACCTC TGGGGTCCCT TCCATCCCGC AGACCCACAG CCCCCAGCAC TGGGAACAAC CCGTCTACAC ACAGCTCACT CGACCTTGAG GAGGCCTCCC ACGAAGGGCG ACGATGGCCG AGATGATCAC ACAGCTCACT CGACCTTGAG GAGGCCTCCC ACGAAGGGCG ACGATGGCCG
AGATGATCCT AAAAATAACC GAAGAAAGAG AGGACCAACC AGAATTCCCT TTGGACATTT
GTGTTTTTTT GTTTTTTAT TTTGTTTTGT TTTTTCTTCT TCTTCTTCTT CCTTAAAGAC
ATTAAGCTA AAGGCAACTC GTACCCAAAT TTCCAAGACA CAAACATGAC CTATCCAAGC
GCATTACCCA CTTGTGGCCA ATCAGTGGCC AGGCCAACCT TGGCTAAATG GAGCAGCGAA
ATCAACGAGA AACTGGACTT TTTAAACCCT CTTCAGAGCA AGCGTGGAG ATGATGGAGA
ATCGTGTGAT CAGTGGCTA AACTCTCTCG CCTGTTTGGA CTTTGTAATT ATTTTTTAGTA
CAGTAATTAA AGAAAAAAGT CCTCTGTGAG GAATATTCTC TATTTTTAGTA TGTACTGTGT ATGATTCATT ACCATTTTGA GGGGATTTAT ACATATTTTT AGATAAAATT AAATGCTCTT ATTTTTCCAA CAGCTAAACT ACTCTTAGTT GAACAGTGTG CCCTAGCTTT TCTTGCAACC AGAGTATTTT TGTACAGATT TGCTTTCTCT TACAAAAAGA AAAAAAAAT CCTGTTGTAT TAACATTTAA AAACAGAATT GTGTTATGTG ATCAGTTTTG GGGGTTAACT TTGCTTAATT CCTCAGGCTT TGCGATTTAA GGAGGAGCTG CCTTAAAAAA AAATAAAGGC CTTATTTTGC AATTATGGGA GTAAACAATA GTCTAGAGAA GCATTTGGTA AGCTTTATGA TATATATT TTTTAAAGAA GAGAAAAACA CCTTGAGCCT TAAAACGGTG CTGCTGGGAA ACATTTGCAC TCTTTTAGTG CATTTCCTCC TGCCTTTGCT TGTTCACTGC AGTCTTAAGA AAGAGGTAAA AGGCAAGCAA AGGAGATGAA ATCTGTTCTG GGAATGTTTC AGCAGCCAAT AAGTGCCCGA GCACACTGCC CCCGGTTGCC TGCCTGGGCC CCATGTGGAA GGCAGATGCC TGCTCGCTCT GTCACCTGTG CCTCTCAGAA CACCAGCAGT TAACCTTCAA GACATTCCAC

fU

TTGCTAAAAT TATTTATTTT GTAAGGAGAG GTTTTAATTA AAACAAAAA AAATTCTTTT TTTTTTTTT TTTTCCAATT TTACCTTCTT TAAAATAGGT TGTTGGAGCT TTCCTCAAAG GGTATGGTCA TCTGTTGTTA. AATTATGTTC TTAACTGTAA CCAGTTTTTT TTTATTTATC TCTTTAATCT TTTTTATTAT TAAAAGCAAG TTTCTTTGTA TTCCTCACCC TAGATTTGTA TAAATGCCTT TTTGTCCATC CCTTTTTTCT TTGTTGTTTT TGTTGAAAAC AAACTGGAAA CTTGTTTCTT TTTTTGTATA AATGAGAGAT TGCAAATGTA GTGTATCACT GAGTCATTTG CAGTGTTTTC TGCCACAGAC CTTTGGGCTG CCTTATATTG TGTGTGTGTG TGGGTGTGTG TGTGTTTTGA CACAAAACA ATGCAAGCAT GTGTCATCCA TATTTCTCTA CATCTTCTCT TGGAGTGAGG GAGGCTACCT GGAGGGGATC AGCCCACTGA CAGACCTTAA TCTTAATTAC TGCTGTGGCT AGAGAGTTTG AGGATTGCTT TTTAAAAAAG ACAGCAAACT TTTTTTTTA TTTAAAAAA GATATATAA CAGTTTTAGA AGTCAGTAGA ATAAAATCTT AAAGCACTCA TAATATGGCA TCCTTCAATT TCTGTATAAA AGCAGATCTT TTTAAAAAAG ATACTTCTGT AACTTAAGAA ACCTGGCATT TAAATCATAT TTTGTCTTTA GGTAAAAGCT TTGGTTTGTG TTCGTGTTTT GTTTGTTTCA CTTGTTTCCC TCCCAGCCCC AAACCTTTTG TTCTCTCCGT ATATACATTG CATTAAAAAG AAA

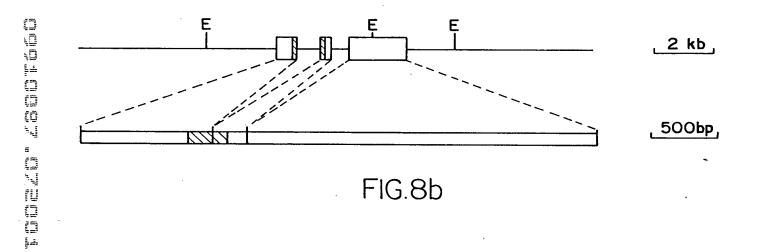
APPROVED	O.G. FIG.			
BY	CLASS	SUBCLASS		
DRAFTSMAN				

Met Asn Leu Leu Asp Pro Phe Met Lys Met Thr Asp Glu Gln Glu Lys Gly Leu Ser Gly Ala Pro Ser Pro Thr Met Ser Glu Asp Ser Ala Gly Ser Pro Cys Pro Ser Gly Ser Gly Ser Asp Thr Glu Asn Thr Arg Pro Gln Glu Asn Thr Phe Pro Lys Gly Glu Pro Asp Leu Lys Lys Glu Ser Glu Glu Asp Lys Phe Pro Val Cys Ile Arg Glu Ala Val Ser Gln Val Leu Lys Gly Tyr Asp Trp Thr Leu Val Pro Met Pro Val Arg Val Asn Gly Ser Ser Lys Asn Lys Pro His Val Lys Arg Pro Met Asn Ala Phe Met Val Trp Ala Gln Ala Ala Arg Arg Lys Leu Ala Asp Gln Tyr Pro His Leu His Asn Ala Glu Leu Ser Lys Thr Leu Gly Lys Leu Trp Arg Leu Leu Asn Glu Ser Glu Lys Arg Pro Phe Val Glu Glu Ala Glu Arg Leu Arg Val Gln His Lys Lys Asp His Pro Asp Tyr Lys Tyr Gln Pro Arg Arg Arg Lys Ser Val Lys Asn Gly Gln Ala Glu Ala Glu Glu Ala Thr Glu Gln Thr His Ile Ser Pro Asn Ala Ile Phe Lys Ala Leu Gln Ala Asp Ser Pro His Ser Ser Ser Gly Met Ser Glu Val His Ser Pro Gly Glu His Ser Gly Gln Ser Gln Gly Pro Pro Thr Pro Pro Thr Thr Pro Lys Thr Asp Val Gln Pro Gly Lys Ala Asp Leu Lys Arg Glu Gly Arg Pro Leu Pro Glu Gly Gly Arg Gln Pro Pro Ile Asp Phe Arg Asp Val Asp Ile Gly Glu Leu Ser Ser Asp Val Ile Ser Asn Ile Glu Thr Phe Asp Val Asn Glu Phe Asp Gln Tyr Leu Pro Pro Asn Gly His Pro Gly Val Pro Ala Thr His Gly Gln Val Thr Tyr Thr Gly Ser Tyr Gly Ile Ser Ser Thr Ala Ala Thr Pro Ala Ser Ala Gly His Val Trp Met Ser Lys Gln Gln Ala Pro Pro Pro Pro Gln Gln Pro Pro Gln Ala Pro Pro Ala Pro Gln Ala Pro Pro Gln Pro Gln Ala Ala Pro Pro Gln Gln Pro Ala Ala Pro Pro Gln Gln Pro Gln Ala His Thr Leu Thr Thr

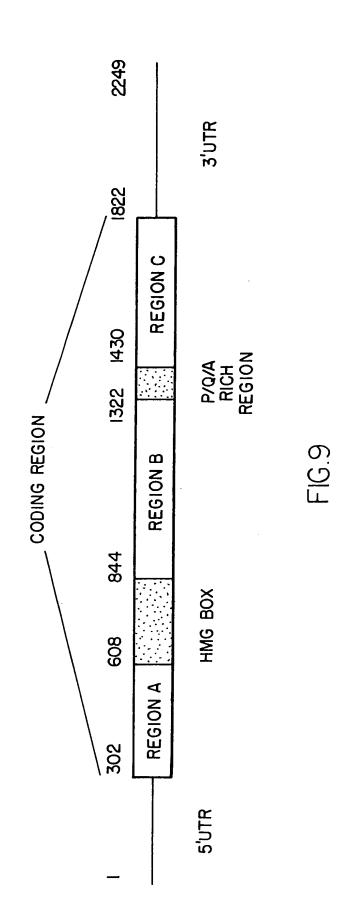
Figure 8a(3)

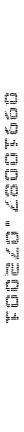
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Gln	Leu	Ser	Pro	Ser 405	His	Tyr	Ser	Glu	Gln 410	Gln	Gln	His	Ser	Pro 415	Gln
	Ile		420					425					430		-
Pro	Pro	Ile 435	Thr	Arg	Ser	Gln	Tyr 440	Asp	Tyr	Thr	Asp	His 445	Gln	Asn	Ser
Ser	Ser 450	Tyr	Tyr	Ser	His	Ala 455	Ala	Gly	Gln	Gly	Thr 460	Gly	Leu	Tyr	Ser
Thr 465	Phe	Thr	Tyr	Met	Asn 470	Pro	Ala	Gln	Arg	Pro 475	Met	Tyr	Thr	Pro	Ile 480
Ala	Asp	Thr	Ser	Gly 485	Val	Pro	Ser	Ile	Pro 490	Gln	Thr	His	Ser	Pro 495	Gln
His	Trp	Glu	Gln 500	Pro	Val	Tyr	Thr	Gln 505	Leu	Thr	Arg	Pro			

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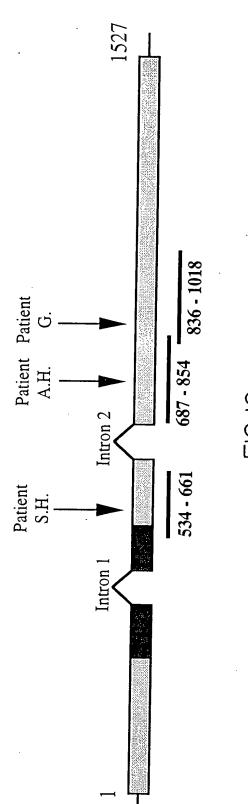


FIG. 10c

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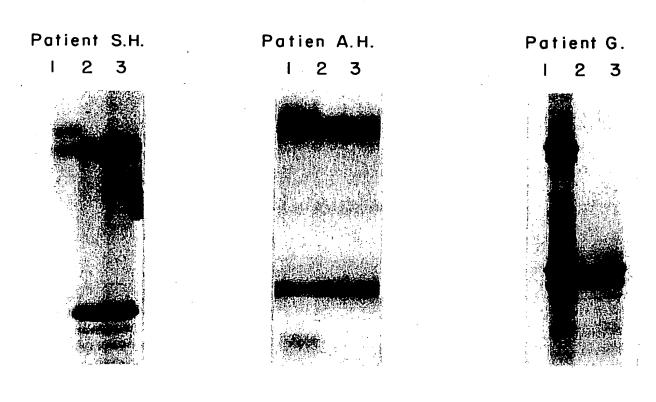


FIG.IOb

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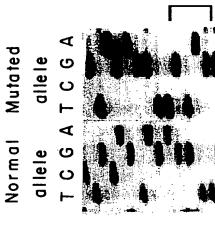
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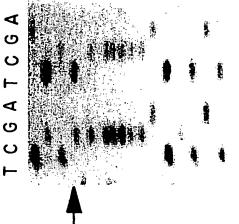
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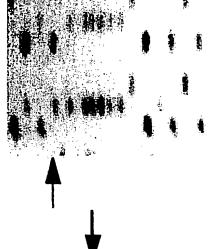
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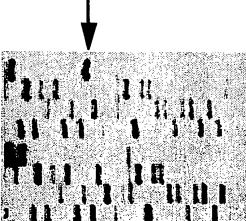
Mutated Normal allele allele

Patient G.









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